



## ring chromosome 20 syndrome

Ring chromosome 20 syndrome is a condition that affects the normal development and function of the brain. The most common feature of this condition is recurrent seizures (epilepsy) in childhood. The seizures may occur during the day or at night during sleep. They are described as partial seizures because they affect only one area of the brain, a region called the frontal lobe. In many cases, the seizures are complex and resistant to treatment with anti-epileptic drugs. Prolonged seizure episodes known as non-convulsive status epilepticus also appear to be characteristic of ring chromosome 20 syndrome. These episodes involve confusion and behavioral changes.

Most people with ring chromosome 20 syndrome also have some degree of intellectual disability and behavioral difficulties. Although these problems can appear either before or after the onset of epilepsy, they tend to worsen after seizures develop. Additional features of this condition can include slow growth and short stature, a small head (microcephaly), and subtle differences in facial features. Major birth defects are rarely seen with ring chromosome 20 syndrome.

### Frequency

Ring chromosome 20 syndrome appears to be a rare condition, although its prevalence is unknown. More than 60 affected individuals have been reported in the medical literature.

### Genetic Changes

Ring chromosome 20 syndrome is caused by a chromosomal abnormality known as a ring chromosome 20 or r(20). A ring chromosome is a circular structure that occurs when a chromosome breaks in two places and its broken ends fuse together. People with ring chromosome 20 syndrome have one copy of this abnormal chromosome in some or all of their cells.

It is not well understood how the ring chromosome causes the signs and symptoms of this syndrome. In some affected individuals, genes near the ends of chromosome 20 are deleted when the ring chromosome forms. Researchers suspect that the loss of these genes may be responsible for epilepsy and other health problems. However, other affected individuals do not have gene deletions associated with the ring chromosome. In these people, the ring chromosome may change the activity of certain genes on chromosome 20, or it may be unable to copy (replicate) itself normally during cell division. Researchers are still working to determine the precise relationship between the ring chromosome 20 and the characteristic features of the syndrome.

## **Inheritance Pattern**

Ring chromosome 20 syndrome is almost never inherited. A ring chromosome typically occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early embryonic development. Often, the ring chromosome is present in only some of a person's cells. This situation is known as mosaicism.

Most affected individuals have no history of the disorder in their families. However, at least one family has been reported in which a ring chromosome 20 was passed from a mother to her children.

## **Other Names for This Condition**

- r(20) syndrome
- ring 20 syndrome
- ring chromosome 20
- ring chromosome 20 epilepsy syndrome

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Ring chromosome 20 syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265482/>

### Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Chromosome  
<https://medlineplus.gov/ency/article/002327.htm>
- MedlinePlus Encyclopedia: Epilepsy  
<https://medlineplus.gov/ency/article/000694.htm>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Encyclopedia: Chromosome  
<https://medlineplus.gov/ency/article/002327.htm>
- Encyclopedia: Epilepsy  
<https://medlineplus.gov/ency/article/000694.htm>
- Health Topic: Epilepsy  
<https://medlineplus.gov/epilepsy.html>

### Genetic and Rare Diseases Information Center

- Ring chromosome 20  
<https://rarediseases.info.nih.gov/diseases/1334/ring-chromosome-20>

### Additional NIH Resources

- National Human Genome Research Institute: Chromosome Abnormalities  
<https://www.genome.gov/11508982/>
- National Institute of Neurological Disorders and Stroke: Epilepsy Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Epilepsy-Information-Page>

### Educational Resources

- Epilepsy Action  
<https://www.epilepsy.org.uk/info/syndromes/ring-chromosome-20-syndrome>
- MalaCards: ring chromosome 20  
[http://www.malacards.org/card/ring\\_chromosome\\_20](http://www.malacards.org/card/ring_chromosome_20)
- March of Dimes: Chromosomal Conditions  
<http://www.marchofdimes.org/baby/chromosomal-conditions.aspx>
- Orphanet: Ring chromosome 20  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=1444](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1444)
- Unique support group booklet about ring chromosome 20  
<http://www.rarechromo.org/information/Chromosome%2020/Ring%2020%20FTNW.pdf>

### Patient Support and Advocacy Resources

- American Epilepsy Society  
<https://www.aesnet.org/>
- Chromosome Disorder Outreach  
<http://chromodisorder.org/>

- Citizens United for Research in Epilepsy (CURE)  
<http://www.cureepilepsy.org/>
- Unique - The Rare Chromosome Disorder Support Group (UK)  
<http://www.rarechromo.org/>

#### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22Ring+Chromosomes%22+OR+%22ring+chromosome+20+syndrome%22>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Ring+Chromosomes%5BMAJR%5D%29+AND+%28%28ring+chromosome+20+syndrome%5BTIAB%5D%29+OR+%28ring+chromosome+20%5BTIAB%5D%29+OR+%28ring+20+syndrome%5BTIAB%5D%29+OR+%28ring+chromosome+20+epilepsy+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

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<http://www.rarechromo.org/information/Chromosome%2020/Ring%2020%20FTNW.pdf>

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